

Dystonia, dyskinesia génpanel v:2025-05-22

Panel tartalma 270 gén:

AAAS, AASS, ABAT, ABCB7, ACBD6, ACER3, ACOX1, ACTB, ACSF3, ADAR, ADCY5, AFG3L2, ALDH18A1, ANO10, ANO3, AOPEP, AP1S2, APTX, ARFGF3, ARSA, ARX, ASL, ATCAY, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP5MC3, ATP7B, ATXN1, ATXN2, ATXN3, AUH, BCAP31, BCS1L, C19orf12, C9orf72, CA8, CACNA1A, CACNA1B, CACNA1G, CACNB4, CAMK4, CHCHD2, CHMP2B, CIZ1, CLN3, CLN5, CLN8, CLPB, COASY, COL6A3, COX10, COX15, COX20, CP, CTSD, CWF19L1, CYP27A1, CSF1R, CSTB, DCAF17, DCC, DCTN1, DDC, DHDDS, DLAT, DLD, DNAJC12, DNAJC5, DNAJC6, ECHS1, EIF2AK2, EIF4G1, ELOVL4, FA2H, FBXO7, FGF14, FITM2, FOLR1, FOXG1, FOXRED1, FTL, FUCA1, FXN, GBA, GCDH, GCH1, GFAP, GJC2, GLB1, GLRA1, GLRB, GM2A, GNAL, GNAO1, GNB1, GRID2, GRIN1, GRM1, GRN, GTPBP2, HCFC1, HECW2, HEXA, HIBCH, HNRNPH1, HPCA, HPRT1, HSD17B10, HSPD1, HTRA2, HTT, IFIH1, IMPDH2, IRF2BPL, ITPR1, JPH3, KCNA1, KCNC3, KCND3, KCNMA1, KCNN2, KCNQ2, KCTD17, KIF1A, KIF1C, KMT2B, L2HGDH, LRPPRC, LRRK2, LYST, MAL, MAPT, MARS2, MECR, MED27, MRE11, MTFMT, MYORG, NDUFA1, NDUFA10, NDUFA12, NDUFA2, NDUFAF5, NDUFAF6, NDUFS1, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NGLY1, NKX2-1, NKX6-2, NPC1, NPC2, NUP54, NUS1, OPA3, PANK2, PARK7, PCCA, PCCB, PCDH12, PDE10A, PDE2A, PDGFB, PDGFRB, PDHA1, PDHX, PDYN, PET100, PINK1, PLA2G6, PLP1, PNKD, PNKP, PNPT1, POLR3A, PPP2R2B, PPP2R5D, PRKCG, PRKN, PRKRA, PRNP, PRRT2, PTS, QDPR, RAB39B, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RNU7-1, SACS, SAMHD1, SCN1A, SCN8A, SERAC1, SETX, SGCE, SHQ1, SIL1, SLC16A2, SLC18A2, SLC19A3, SLC1A3, SLC20A2, SLC2A1, SLC30A10, SLC30A9, SLC39A14, SLC6A3, SLC6A5, SLC6A8, SNCA, SNORD118, SNX14, SPATA5L1, SPG11, SPG7, SPR, SQSTM1, STUB1, SUCLA2, SUOX, SURF1, SYNJ1, SYT1, TAF1, TARS2, TBC1D24, TBK1, TBP, TGM6, TH, THAP1, TIMM8A, TMEM151A, TMEM240, TOR1A, TPK1, TPP1, TREX1, TSPOAP1, TTBK2, TUBB4A, UBTF, UCHL1, VAC14, VAMP1, VAMP2, VPS11, VPS13A, VPS13D, VPS16, VPS35, VPS41, VPS4A, WDR45, WDR73, WFS1, WWOX, XK, XPR1, YIF1B, YY1, ZSWIM6

Forrás:

Dystonia - PS128100

<https://omim.org/phenotypicSeries/PS128100>

Episodic kinesigenic dyskinesia - PS128200

<https://omim.org/phenotypicSeries/PS128200>

Early onset dystonia (Version 1.149)

<https://panelapp.genomicsengland.co.uk/panels/192/>

Childhood onset dystonia, chorea or related movement disorder (Version 7.1)

<https://panelapp.genomicsengland.co.uk/panels/847/>

Adult onset dystonia, chorea or related movement disorder (Version 5.1)

<https://panelapp.genomicsengland.co.uk/panels/540/>

(Megtekintve: 2025-05-22)